THE KAPIʻOLANI MEDICAL CENTER FOR WOMEN AND CHILDREN
HONOLULU, HAWAII

CALIFORNIA DEPARTMENT OF HEALTH SERVICES, GENETIC DISEASE BRANCH
BERKELEY, CALIFORNIA

INFORMED CONSENT TO TAKE PART IN A CLINICAL RESEARCH STUDY

Title of Study: Neonatal Screening for Inborn Errors of Metabolism Using Tandem Mass Spectrometry (MS/MS)

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741 Sunset Avenue, Honolulu, HI 96816

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California Department of Health Services, Genetic Disease Branch
2151 Berkeley Way, Annex 4, Berkeley, CA 94704

INFORMED CONSENT

I have been asked to take part in this research study. Before I decide to take part, I will read the following information carefully and ask questions so that I fully understand the requirements.

Before I decide whether or not to take part in this study, I must understand the purpose, how it may help, any risks to me, and what I have to do. This process is called informed consent. The consent form gives me information about the study, which will be discussed with me. Once I understand the study, and if I agree to take part, I will be asked to sign this consent form. I will be given a copy to keep.

Before I learn about the study, it is important that I know the following:

- Taking part in this study is of my own free will.
- I may decide not to take part in the study or stop being in the study at any time without it making any difference to my baby's care now or in the future, or to any benefits that my baby is allowed.
- If the study changes in any way that could make a difference to my taking part, I will be told about the changes and may be asked to sign a new consent form.

PURPOSE OF THE STUDY

The purpose of this pilot research study is to evaluate testing methods for identifying over twenty additional metabolic disorders that are currently not part of the mandated newborn screening testing. This supplemental testing, using tandem mass spectrometry, will be done after the required newborn screening testing. Currently, five blood spots are collected for the required newborn screening testing of seven disorders. The supplemental testing will use the same blood specimen that has already been drawn for the required newborn screening tests. No additional blood spots will be needed.

WHO MAY TAKE PART IN THE STUDY

All newborns in Hawaii, born between March 1, 2002 to December 31, 2002, may take part in the study. It is anticipated that 2,300 newborns may be participating in this research project.

PROCEDURES

You will receive information about the supplemental screening of over twenty additional disorders from trained personnel. You will be asked to sign an informed consent form if you would like your baby to receive the supplemental screening.

RISKS

Your baby will experience no additional physical discomfort beyond the heelstick normally done for routine newborn screening. There is a risk for pain, bleeding, or bruising at the place where the lancet enters the heel. Risk of infection is minimal because only sterile one-time use lancets will be used by trained personnel.

Information gained during this study and information known about my baby will be kept confidential (private) to the extent permitted by state and federal laws and will not given to anyone without my written consent (permission).

Reporting of unusual results for supplemental testing may be delayed because the mandated newborn screening testing is done first. However, your baby's physician will be notified if the specimen is unsuitable for supplemental testing or if there are positive or unusual results requiring further follow-up. Your baby's physician will discuss these results with you. If you are contacted, your baby will be referred to a doctor who specializes in the treatment of metabolic disorders. A positive or an unusual result does not always mean that a disorder is present. There is also a small chance that these rare disorders could be missed by this test. If the specimen collected from the baby is found to be unsuitable for testing, the supplemental testing will not be done.

BENEFITS

The supplemental testing of over twenty additional disorders detected by tandem mass screening, may result in earlier detection and treatment, thus preventing serious physical and mental disabilities, and possibly, even death. There could be disorders identified by the study which may not cause significant health problems or require treatment. There may also be disorders identified for which there is no effective treatment.

In addition, there is potential benefit to the greater community and to science for the knowledge obtained during the pilot study in terms of setting appropriate cutoffs for detection; determining which abnormal screens are clinically significant; determining which of the rarer disorders may have improved outcomes or reduced costs of care from the early detection and treatment; and obtaining greater knowledge of the frequency of these disorders in the general population.

By participating in the study, you are helping the Hawaii Newborn Metabolic Screening Program to decide which disorders to add to mandated newborn screening in the future.

COSTS

There will be no cost to you for the supplemental screening. If confirmatory testing is needed, these laboratory costs will be billed to the Department of Health Newborn Metabolic Screening Program.

COMPENSATION

There will be no compensation to participate in this research project. If a disorder is diagnosed, the cost of the treatment will be the responsibility of the family.

CONFIDENTIALITY

If you decide to participate in this research project, your physician will be notified that your baby's specimen was sent to California for supplemental testing. Participation in this research project also means consenting to the release of your baby's demographic information (for example, sex, birth date, birth weight, ethnicity), necessary for supplemental testing and follow-up, to the Hawaii Newborn Metabolic Screening Program. You might be contacted directly for some of this information if it is not available elsewhere. All information collected shall be confidential and shall not be released to anyone without your written permission.

VOLUNTARY PARTICIPATION

Your participation in this research project is voluntary, and if you decide not to participate at any time, you will not lose any existing benefits or services. Your baby will still received mandated newborn screening.

ALTERNATIVES

If you do not wish to participate in this project but wish to have supplemental screening through a private laboratory, your doctor, the hospital staff, or the Newborn Screening Program can assist you. There is a cost to you for supplemental screening through a private laboratory.

NEW FINDINGS

Any important new information discovered during this study that may make a difference in your willingness to continue in the research study will be given to you.

PARTIES TO CONTACT

Contact the investigators listed on the first page of the consent form if you have any questions about the research study. Any other questions that you have about your rights as a participant should be directed to any of the following:

Hawaii Pacific Health Institutional Review Board
Raul Rudoy, M.D., Chair
55 Merchant St., 27th Floor
Honolulu, HI 96813
Phone: (808) 535-7500

State of Hawaii Department of Health
Human Research Assistance and Evaluation Committee
Bart Aronoff, M.P.H., Executive Secretary, HRAEC
P.O. Box 3378
Honolulu, HI 96801-3378
Phone: (808) 586-4580

UNDERSTANDING AND COMPLIANCE

My signature below indicates that I have read and that I understand the procedures described above and give my informed and voluntary consent to have my child or grandchild participate in this study. I understand that I will receive a signed copy of this consent form.

_____	_____
Child's Name	Birth Date
_____	_____
Parent or Legal Guardian Signature	Date
_____	_____
Witness Signature	Date